# Nearsightedness

Nearsightedness, also known as myopia, is an eye condition that causes blurry distance vision. People who are nearsighted have more trouble seeing things that are far away (such as when driving) than things that are close up (such as when reading or using a computer). If it is not treated with corrective lenses or surgery, nearsightedness can lead to squinting, eyestrain, headaches, and significant visual impairment.

Nearsightedness usually begins in childhood or adolescence. It tends to worsen with age until adulthood, when it may stop getting worse (stabilize). In some people, nearsightedness improves in later adulthood.

For normal vision, light passes through the clear cornea at the front of the eye and is focused by the lens onto the surface of the retina, which is the lining of the back of the eye that contains light-sensing cells. People who are nearsighted typically have eyeballs that are too long from front to back. As a result, light entering the eye is focused too far forward, in front of the retina instead of on its surface. It is this change that causes distant objects to appear blurry. The longer the eyeball is, the farther forward light rays will be focused and the more severely nearsighted a person will be.

Nearsightedness is measured by how powerful a lens must be to correct it. The standard unit of lens power is called a diopter. Negative (minus) powered lenses are used to correct nearsightedness. The more severe a person's nearsightedness, the larger the number of diopters required for correction. In an individual with nearsightedness, one eye may be more nearsighted than the other.

Eye doctors often refer to nearsightedness less than -5 or -6 diopters as "common myopia." Nearsightedness of -6 diopters or more is commonly called "high myopia." This distinction is important because high myopia increases a person's risk of developing other eye problems that can lead to permanent vision loss or blindness. These problems include tearing and detachment of the retina, clouding of the lens (cataract), and an eye disease called glaucoma that is usually related to increased pressure within the eye. The risk of these other eye problems increases with the severity of the nearsightedness. The term "pathological myopia" is used to describe cases in which high myopia leads to tissue damage within the eye.

# Frequency

Nearsightedness is the most frequent cause of correctable visual impairment worldwide, and it has become increasingly common over the past few decades. By 2020, scientists estimate that more than one-third of the world population, about 2.6 billion people, will have myopia. Almost 400 million of those will have high myopia.

The prevalence of nearsightedness is significantly higher in some East Asian countries, where the condition affects up to 90 percent of young adults. Most of these individuals have common myopia. However, in regions where myopia is most common, between 10 and 20 percent of young adults have high myopia.

#### Causes

Nearsightedness is typically a complex condition. Multiple genetic variations, each with a small effect, likely interact with environmental and lifestyle factors to influence whether a person becomes nearsighted. Some of the factors that contribute to nearsightedness have been confirmed by research, while others have yet to be discovered.

Occasionally, nearsightedness (particularly high myopia) results from mutations in a single gene. Variations in at least seven specific genes have been associated with high myopia. In some families, the genetic cause of their nearsightedness has been narrowed down to a small segment of a chromosome (called a locus, plural loci). Each locus can contain dozens or hundreds of genes; researchers have not determined which genes are involved. More than two dozen loci related to nearsightedness have been identified. Each one is named with the prefix "MYP" (for "myopia") and a number that reflects the order in which it was reported.

Large studies have identified more than 200 genes involved in nearsightedness, and additional studies are underway. Some of these genes help guide eye growth before and after birth. Other genes are involved in processing light signals in the retina. Still other genes are known to be involved in nearsightedness, but their role in vision is unclear. Environmental and lifestyle factors also play an important part in nearsightedness. Much of the recent increase in the frequency of nearsightedness worldwide is likely related to spending less time outdoors and doing more "near work," such as reading, studying, and working on computers and handheld devices. Researchers are working to determine how genetic variations may interact with these lifestyle changes to alter the shape of the eyes.

In most nearsighted people, this vision problem is not part of a larger genetic syndrome. However, more than 200 genetic conditions, most of them rare, include nearsightedness as a feature. These conditions include autosomal recessive congenital stationary night blindness, X-linked congenital stationary night blindness, Stickler syndrome, Marfan syndrome, retinitis pigmentosa, cone-rod dystrophy, deafness and myopia syndrome, Knobloch syndrome, and Cohen syndrome.

#### Inheritance Pattern

Because common myopia is a complex condition involving hundreds of genes, the condition does not have a clear pattern of inheritance. The risk of developing this condition is greater for first-degree relatives of affected individuals (such as siblings or children) as compared to the general public. This increased risk is likely due in part to

shared genetic factors, but it may also be related to environment and lifestyle factors that are shared by members of a family.

Like common myopia, high myopia seldom has a clear pattern of inheritance. However, when it is caused by mutations in a single gene, it can follow an autosomal dominant, autosomal recessive, or X-linked inheritance pattern. In autosomal dominant inheritance, one copy of an altered gene in each cell is sufficient to cause the disorder. In many cases, an affected person inherits the gene mutation from an affected parent. In autosomal recessive inheritance, both copies of a gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. X-linked inheritance applies to mutations in genes located on the X chromosome, one of the two sex chromosomes in each cell. In males, who have only one X chromosome, a mutation in the only copy of the gene in each cell is sufficient to cause the condition. In females, who have two copies of the X chromosome, one altered copy of the gene in each cell can lead to less severe features of the condition or may cause no signs or symptoms at all. In general, males are affected by X-linked disorders much more frequently than females.

When nearsightedness is a feature of a genetic syndrome, it follows the inheritance pattern of that syndrome, most commonly autosomal dominant, autosomal recessive, or X-linked.

#### Other Names for This Condition

- close sighted
- myopia
- myopic
- near-sightedness
- nearsighted
- short-sighted
- short-sightedness

#### **Diagnosis & Management**

# Formal Treatment/Management Guidelines

 American Optometric Association: Optometric Clinical Practice Guideline: Care of the Patient with Myopia https://www.aoa.org/documents/optometrists/CPG-15.pdf

# **Genetic Testing Information**

- What is genetic testing?
   /primer/testing/genetictesting
- Genetic Testing Registry: Myopia https://www.ncbi.nlm.nih.gov/gtr/conditions/C0027092/

# Research Studies from ClinicalTrials.gov

ClinicalTrials.gov
 https://clinicaltrials.gov/ct2/results?cond=%22nearsightedness%22+OR+%22Myopia%22

## Other Diagnosis and Management Resources

- American Academy of Ophthalmology: Myopia Diagnosis https://www.aao.org/eye-health/diseases/myopia-nearsightedness-diagnosis
- American Academy of Ophthalmology: Myopia Treatment https://www.aao.org/eye-health/diseases/myopia-nearsightedness-treatment
- American Optometric Association: Find a Doctor https://www.aoa.org/doctor-locator-search
- American Optometric Association: Quick Reference Guide: Care of the Patient with Myopia https://www.aoa.org/documents/optometrists/QRG-15.pdf
- MedlinePlus Medical Tests: Vision Screening https://medlineplus.gov/lab-tests/vision-screening/

#### **Additional Information & Resources**

#### Health Information from MedlinePlus

- Encyclopedia: Nearsightedness https://medlineplus.gov/ency/article/001023.htm
- Health Topic: Refractive Errors https://medlineplus.gov/refractiveerrors.html
- Medical Tests: Vision Screening https://medlineplus.gov/lab-tests/vision-screening/

#### Genetic and Rare Diseases Information Center

Myopia 6
 https://rarediseases.info.nih.gov/diseases/9937/myopia-6

#### Additional NIH Resources

- National Eye Institute: Facts About Myopia https://nei.nih.gov/health/errors/myopia
- NIH News in Health: A Blurry Worldview: Understanding Myopia https://newsinhealth.nih.gov/2016/07/blurry-worldview

#### **Educational Resources**

- American Academy of Ophthalmology https://www.aao.org/eye-health/diseases/myopia-nearsightedness
- American Optometric Association https://www.aoa.org/patients-and-public/eye-and-vision-problems/glossary-of-eye-and-vision-conditions/myopia
- healthychildren.org from the American Academy of Pediatrics https://www.healthychildren.org/English/health-issues/conditions/eyes/Pages/ Myopia-Nearsightedness.aspx
- KidsHealth from the Nemours Foundation: Your Child's Vision https://kidshealth.org/en/parents/vision.html
- MalaCards: myopia https://www.malacards.org/card/myopia
- NHS Choices (UK) https://www.nhs.uk/conditions/short-sightedness/
- Orphanet: OBSOLETE: Syndromic myopia https://www.orpha.net/consor/cgi-bin/OC\_Exp.php?Lng=EN&Expert=98620
- Stanford at The Tech: Genetics of Eyesight https://genetics.thetech.org/ask/ask73
- University of Michigan Kellogg Eye Center https://www.umkelloggeye.org/conditions-treatments/myopia-nearsightedness

### Patient Support and Advocacy Resources

- Low Vision Gateway http://www.lowvision.org/
- Prevent Blindness
   https://www.preventblindness.org/
- Resource List from the University of Kansas Medical Center: Blind/Visual Impairment http://www.kumc.edu/gec/support/visual.html

#### Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28myopia%5BMAJR%5D%29+AND+%28%28gene%5BTIAB%5D%29+OR+%28genes%5BTIAB%5D%29+OR+%28genetic\*%5BTIAB%5D%29%29+AND+%28%28nearsighted\*%5BTI%5D%29+OR+%28myop\*%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

## Catalog of Genes and Diseases from OMIM

- MYOPIA 1, X-LINKED http://omim.org/entry/310460
- MYOPIA 2, AUTOSOMAL DOMINANT http://omim.org/entry/160700
- MYOPIA 3, AUTOSOMAL DOMINANT http://omim.org/entry/603221
- MYOPIA 5, AUTOSOMAL DOMINANT http://omim.org/entry/608474
- MYOPIA 6 http://omim.org/entry/608908
- MYOPIA 7 http://omim.org/entry/609256
- MYOPIA 8 http://omim.org/entry/609257
- MYOPIA 9 http://omim.org/entry/609258
- MYOPIA 10 http://omim.org/entry/609259
- MYOPIA 11, AUTOSOMAL DOMINANT http://omim.org/entry/609994
- MYOPIA 12, AUTOSOMAL DOMINANT http://omim.org/entry/609995
- MYOPIA 13, X-LINKED http://omim.org/entry/300613
- MYOPIA 14 http://omim.org/entry/610320
- MYOPIA 15, AUTOSOMAL DOMINANT http://omim.org/entry/612717

- MYOPIA 16, AUTOSOMAL DOMINANT http://omim.org/entry/612554
- MYOPIA 17, AUTOSOMAL DOMINANT http://omim.org/entry/608367
- MYOPIA 18, AUTOSOMAL RECESSIVE http://omim.org/entry/255500
- MYOPIA 19, AUTOSOMAL DOMINANT http://omim.org/entry/613969
- MYOPIA 20, AUTOSOMAL DOMINANT http://omim.org/entry/614166

#### Medical Genetics Database from MedGen

 Myopia https://www.ncbi.nlm.nih.gov/medgen/44558

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